

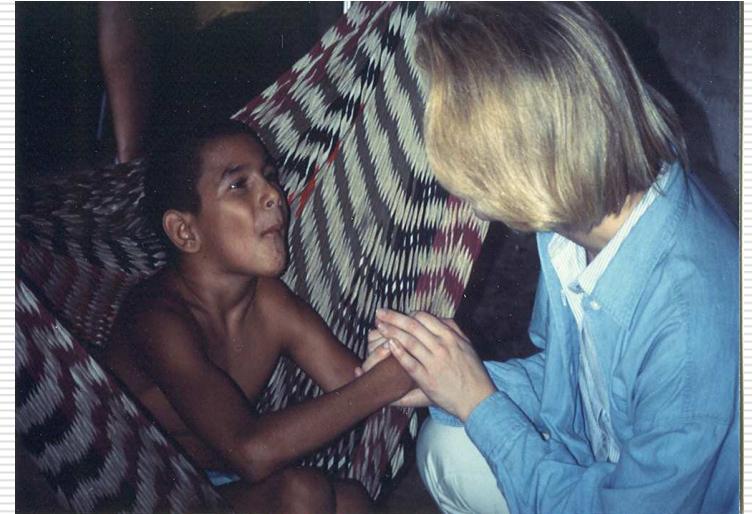
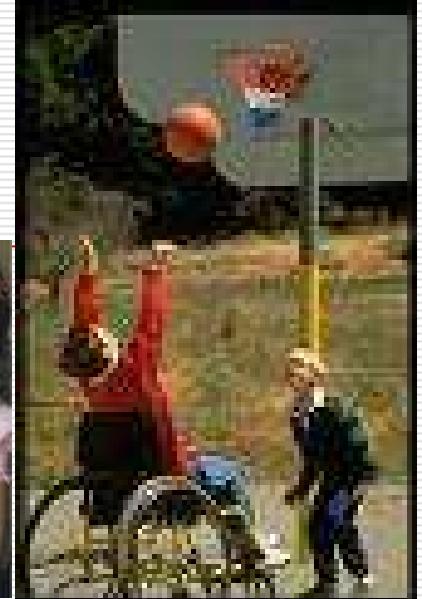
# Juvenile Onset Huntington's Disease

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HDSA Annual Convention  
2009

Jane S. Paulsen

With input from:  
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Helen Brewer  
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## Juvenile HD

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- HD age of onset 10% < 20; 25% >50
- Male transmission 70-80% JuvHD
- CAG > 60 considered most common;  
46% < 60 (42, 45, ...) >80 repeats  
rare (<5% JuvHD); largest CAG=250
- Avg range before diagnosis 9y (0-21)
- 66% report psychiatric/behavioral 1st
- 80% have psychiatric disturbance

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# INTERESTING FAMILY

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- Father: onset age 35 with 47 CAG repeats
- Daughter 1: onset age 4 with 99 CAG repeats
- Daughter 2: onset age 12 with 47 CAG repeats
- Daughter 3: onset age 15 with 47 CAG repeats
- Daughter 1 fits classical definition of paternal anticipation, but daughters 2 + 3 do not (other genes?, childhood onset of adult HD?)

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# Large Differences in JHD

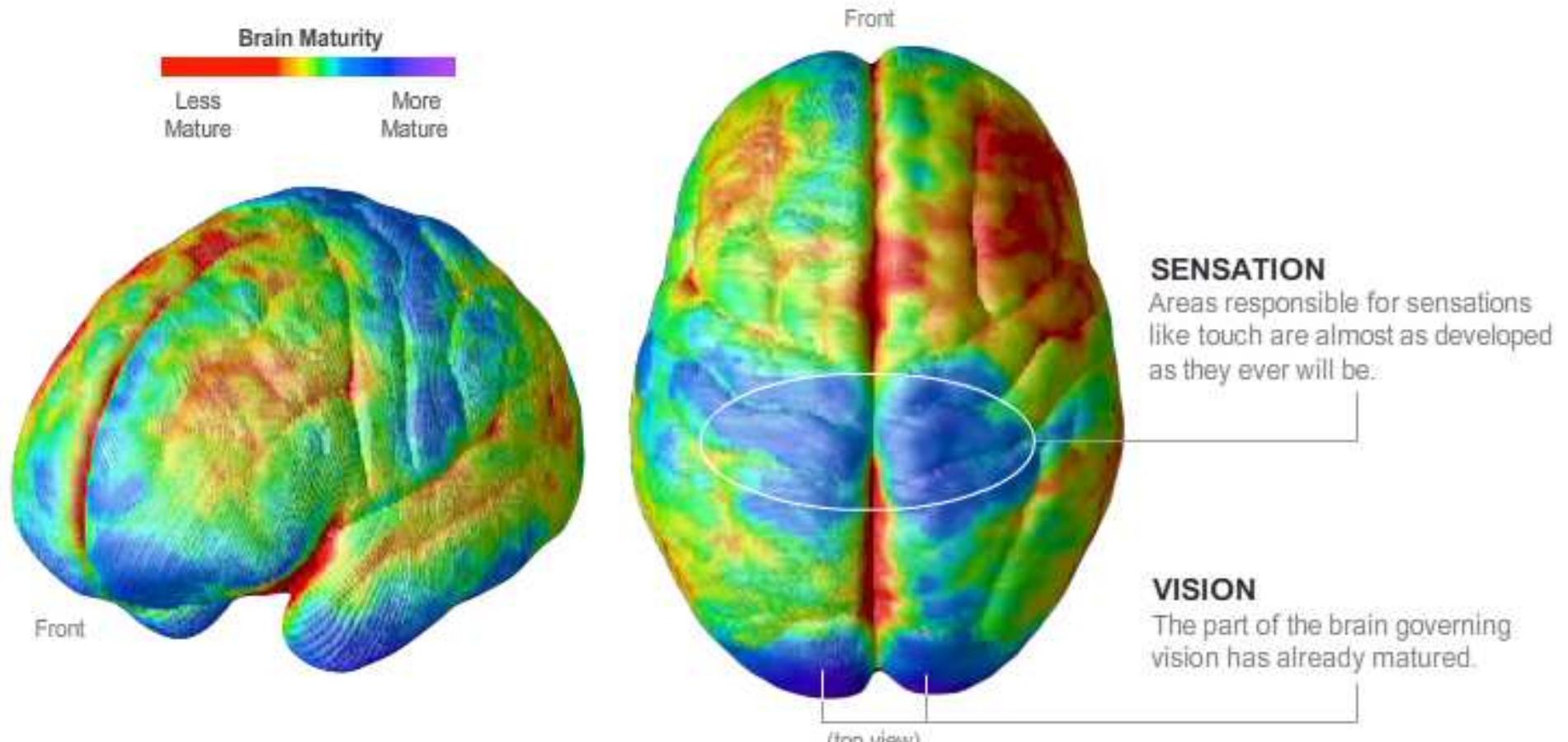
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## □ Children:

- 1. selective vulnerability in children different from adults
- 2. arrest of certain brain regions differs with age.
- 3. degeneration occurs in arrested brain regions.
- 4. unlike adults, compensation of dysfunction is limited by immature brain.

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# Brain Development



# **Motor presentation in Juv HD**

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- Family history positive/confirmatory for HD**
- Stiffness of arms and legs: rigidity and dystonia**
- Clumsiness of arms and legs**
- Seizures**
- Bradykinesia rather than chorea**

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# HD PRESENTATIONS IN CHILDREN FROM CLINICAL CASES: Reynolds

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- RIGID-WESTPHAL JHD VARIANT
- MYOCLONIC JHD VARIANT W CORTICAL ATROPHY
- DYSTONIA JHD VARIANT W CORTICAL ATROPHY
- ATAXIA JHD VARIANT W SEIZURES
- ATAXIA JHD VARIANT W HYPERACTIVITY/ADD
- ATAXIA JHD VARIANT W CORICAL ATROPHY
- DELUSIONAL/OCD JHD VARIANT
- CHILDHOOD ONSET ADULT PATTERN (JHD?)
- PRECLINICAL JHD (CAG'S > 60)
- PRECLINICAL ADULT HD (CAG'S < 60)

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# JHD SCALE

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- 1. Body Language (0-3)
- 2. Eye Movements (0-3)
- 3. Sialorrhea (0-3)
- 4. Ataxia (0-3)
- 5. Dyskinesias (0-3)
- 6. Manual Incoordination (0-3)
- 7. Hallucinations (0-3)
- 8. Compulsive/Obsessive Behavior (0-3)
- 9. Fluctuations In Abnormal Behavior (0-3)

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# Cognitive symptoms

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- Trouble learning new things
- Inattention, less concentration
- Difficulty starting or completing a task
- Difficulty doing things in a sequence
- Regression or losses can be seen in many areas previously attained; request evaluations of standardized test performances

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# **Cognitive interventions**

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- Individualized Education Plan for school aged, IFSP for young children (both mandated by IDEA legislation)
- Ongoing adjustments to home and school environments to allow adaptation to brain changes and their consequences
- School-home partnership (or child care)

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# **Psychiatric and Behavioral Symptoms**

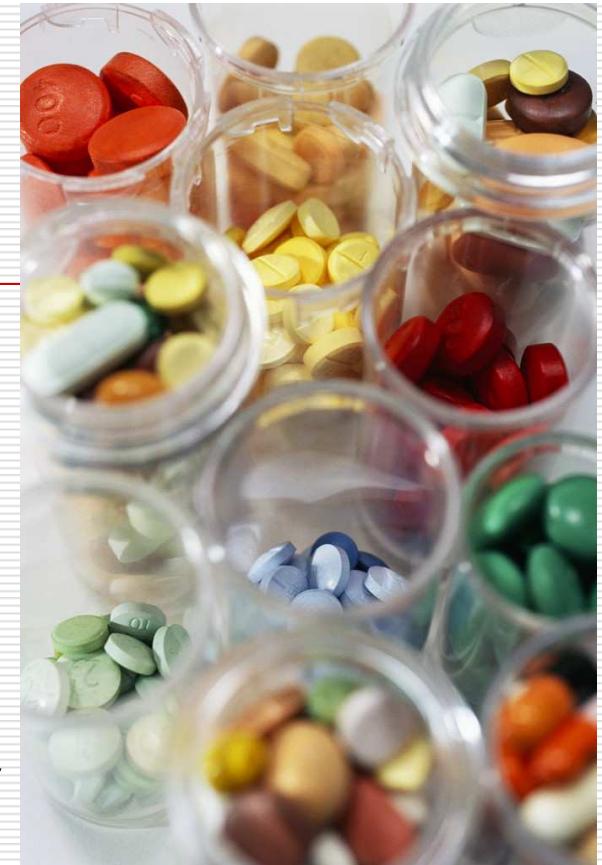
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- Depression is most common symptom in children
- Sad affect, apathy, withdrawal, tearfulness, changes in sleep and appetite
- Suicide risk needs to be assessed
- May manifest as irritability in teens
- May be bipolar or cyclothymic

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# Medications

- Antidepressants
  - Tricyclics
  - SSRI
  - SNRI
- Mood stabilizers
- Beta-andrenergics, sedatives, neuroleptics for aggression
- Antipsychotics
- Anticonvulsants for seizures
- Treatments for chorea
- Treatments for spasticity/rigidity/dystonia



# Non-medicine treatments

- Physical/Occupational therapy
- Speech therapy
- Assistive devices
- Safety evaluation
- Counseling
- Behavior management
- Independent educational plan
- Alternative therapies
- Professional support...hygiene coach
- Respite care



# Efforts to enrich and normalize...



racing



dancing



skating



painting

# Primary Principles: Behavior Management in Children

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- Consider time perception and age
- Pick your battles
- Planning and Routine
- Intentional and Unintentional reinforcement
- Follow-through

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# Behavior Management Strategies

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- ABC: Antecedent, Behavior, Consequences
- Parent Effectiveness Training
- Collaborative Problem-Solving
- Russell Barkley, Ross Greene

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# Prevention of behavior through anticipation

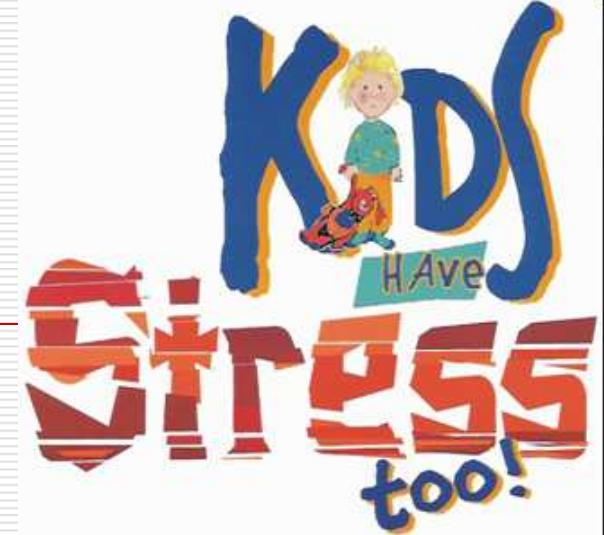
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- Routines
- Clear structure and rituals
- Use cues via timer or voice prior to transitions
- Offer controlled choices
- Reward positive choices
- Identify common and recurring triggers
- Focus on major and important areas of conflict

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# Factors affecting behavior

- Frustration
  - Task complexity
  - Organizational need
  - Restraint needed for setting
  - Communication or other limitations
- Competition
  - Stimulation level in environment
  - Evident or presumed alternatives
- Fatigue
  - Time of day
  - Wellness
  - Tiredness

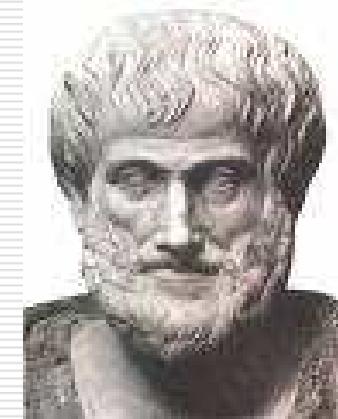


# Anger

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“Anyone can become angry, that is easy; but to be angry with the right person, to the right degree, at the right time, for the right purpose, and in the right way...this is not easy.”

Aristotle



# Outbursts, temper...

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- Identify the antecedent or “trigger”
  - Bath example

- Choose your basket

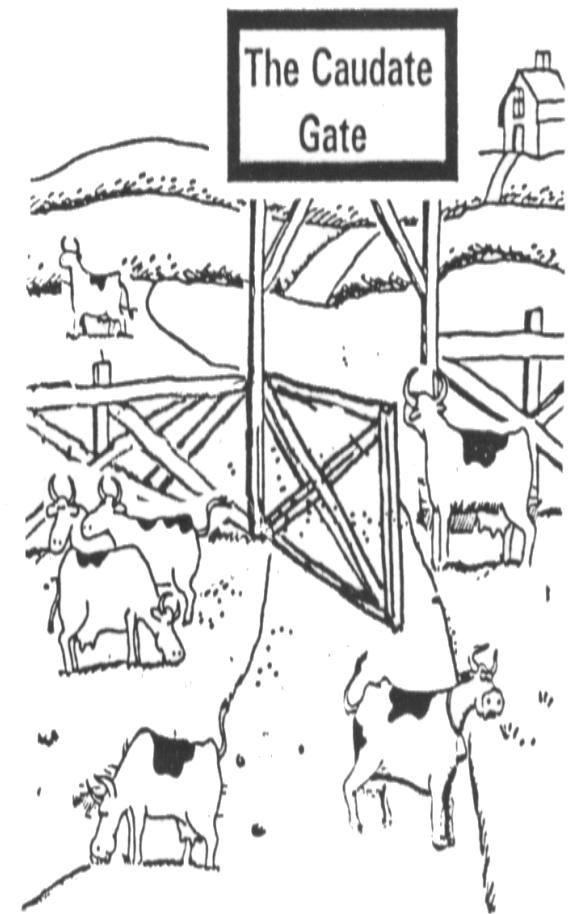
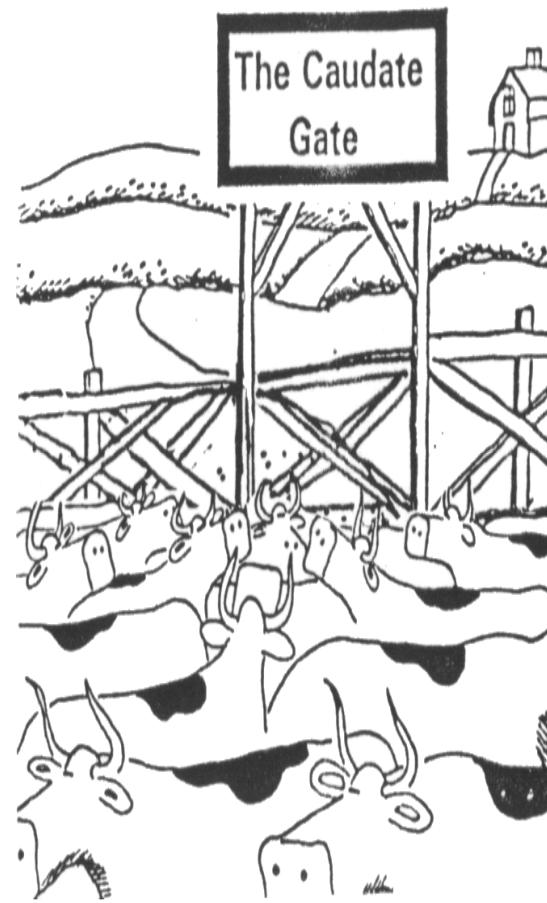
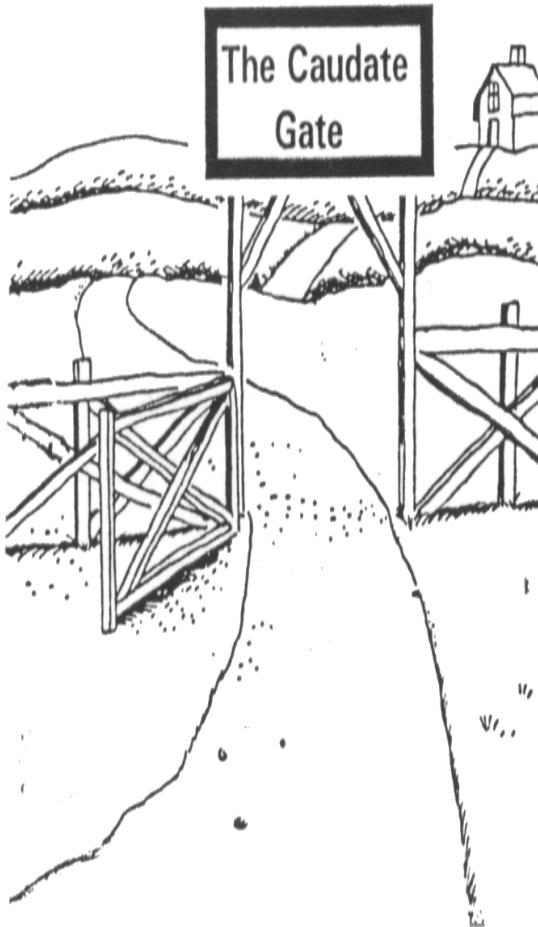
- Basket A – safety
  - Basket B – priority
  - Basket C – what or how does it really matter?



- Acknowledge FEELINGS
- Assure safe environment(s)
- Check factors affecting behavior (FAB)

# “Gates” in the Brain

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# What Happens When Gates Fall Out of Repair?

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- Poor control of temper
- Poor control of recall
- Dysregulation of emotion and thinking
- Confusion



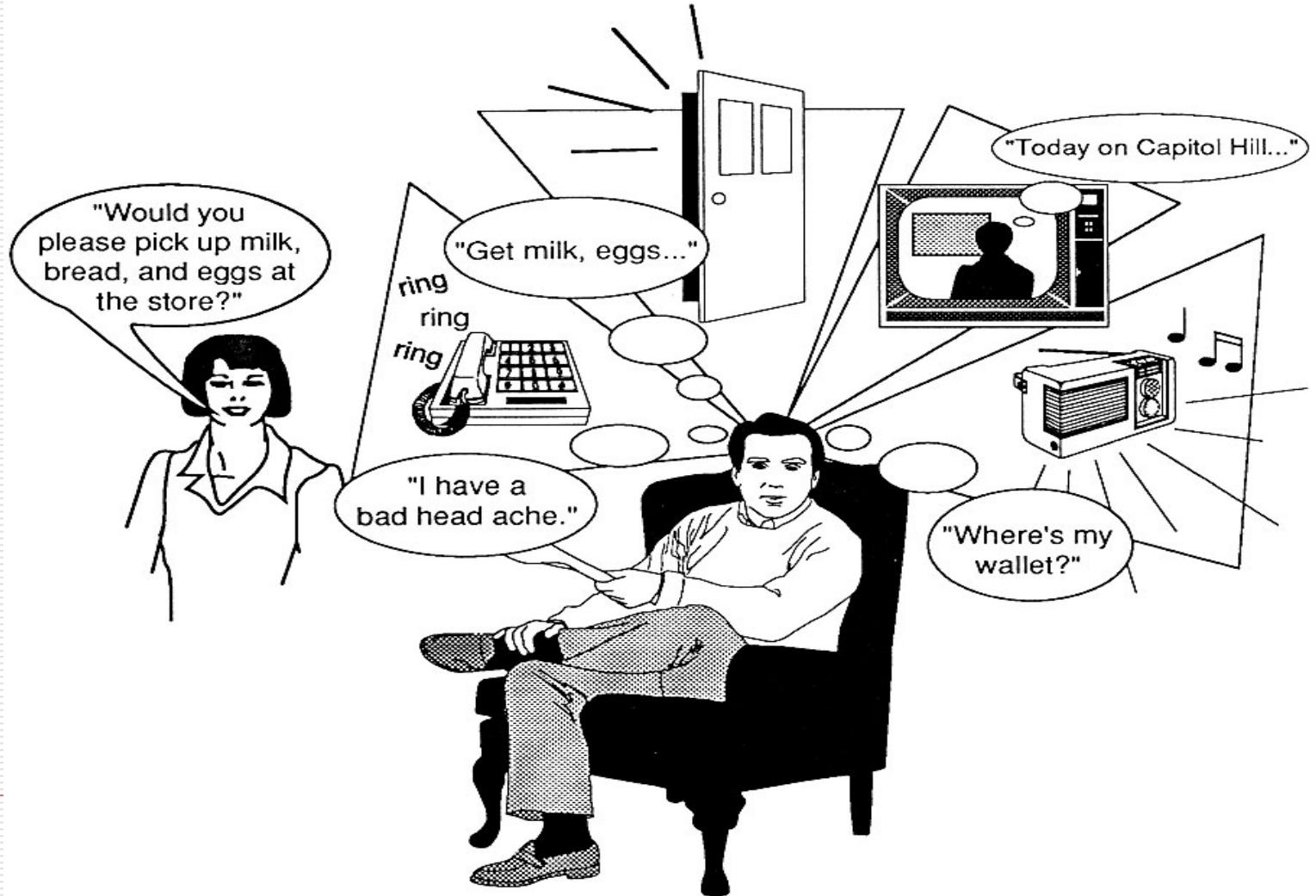
# Inhibition: Being able to choose behavior

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- Disease produces disinhibition of impulses
- Sexualized behavior can be poorly filtered
- Violence and aggression, either verbal or physical
- May lead to legal difficulties
- Can also lead to substance use in adolescents

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# Filtering, organizing, prioritizing



# Addressing attention and “on-task” behavior

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- Establish routine
- Baby steps
- Modify requests to parts that can be attained
- Celebrate accomplishments
- Reinforce positively – go for it!



# **Obsessiveness**

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- May be very slow with beginning tasks or completing things due to rituals or compulsions
- Perseveration in thoughts, words or action
- May become very angry when disrupted or asked to deviate from the usual pattern

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# Managing Obsessions

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- Allow child to talk about interests
- Be creative integrating interests into school and home activities
- Gently shift to new topic or activity
- Recognize as symptom, nor purposeful or willful
- Use prompts frequently to cue shift in activity or topic
- Agree to return to desired topic after “x”

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# Sexuality

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- Changes of puberty may require acquisition of new personal care and social skills
- Inability to plan and direct attention
- May be exhibited in inappropriate settings, i.e. public masturbation
- May result in promiscuity or sexual abuse
- Can become victims or perpetrators of sexual assault

# Team Approach

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- Social services
- Financial planning
- School programs
- Physical supports
- Medical symptom treatments
- Nutrition/exercise schedules
- Behavior Management Skills
- Respite and Support for all Well-Being



# Maximizing wellness and slowed progression

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- Enriched environment
- Low stress
- Nutrition
- Exercise
- Wellness of caregiver
- Wellness of family



# The rest of the family

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- Take breaks
- Tag-team
- Allow change and flexibility
- Breathe
- Forgive yourself and others
- Each day is a new day

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